

Chapter 14: Mendel and the Gene Idea

If you have completed a first-year high school biology course, some of this chapter will serve as a review for the basic concepts of Mendelian genetics. For other students, this may be your first exposure to genetics. In either case, this is a chapter that should be carefully mastered. Spending some time with this chapter, especially working genetics problems, will give you a solid foundation for the extensive genetics unit in the chapters to come.

Overview:

1. In the 1800s the most widely favored explanation of genetics was “blending.” Explain the concept of blending, and then describe how Mendel’s “particulate” (gene) hypothesis was different.

The explanation of heredity most widely in favor during the 1800s was the “blending” hypothesis, the idea that genetic material contributed by the two parents mixes in a manner analogous to the way blue and yellow paints blend to make green. This hypothesis predicts that over many generations, a freely mating population will give rise to a uniform population of individuals. However, our everyday observations and the results of breeding experiments with animals and plants, contradict that prediction. The blending hypothesis also fails to explain other phenomena of inheritance, such as traits reappearing after skipping a generation. An alternative to the blending model is a “particulate” hypothesis of inheritance: the gene idea. According to this model, parents pass on discrete heritable units—genes—that retain their separate identities in offspring.

Concept 14.1 Mendel used the scientific approach to identify two laws of inheritance

2. One of the keys to success for Mendel was his selection of pea plants. Explain how using pea plants allowed Mendel to control mating; that is, how did this approach let Mendel be positive about the exact characteristics of each parent?

The reproductive organs of a pea plant are in its flowers, and each pea flower has both pollen-producing organs (stamens) and an egg-bearing organ (carpel). In nature, pea plants usually self-fertilize: Pollen grains from the stamens land on the carpel of the same flower, and sperm released from the pollen grains fertilize eggs present in the carpel. To achieve cross-pollination (fertilization between different plants), Mendel removed the immature stamens of a plant before they produced pollen and then dusted pollen from another plant onto the altered flowers. Each resulting zygote then developed into a plant embryo encased in a seed (pea). Mendel could thus always be sure of the parentage of new seeds.

3. What is the difference between a *character* and a *trait*? Explain using an example.

A heritable feature that varies among individuals, such as flower color, is called a character. Each variant for a character, such as purple or white color for flowers, is called a trait. For example, the varying color of the flowers on pea plants is a character, and the specific variations, white and purple, are traits.

4. Define the following terms. Then, consider your own family. Which generation would your mother's grandparents be? Your mother? You?

P generation: Parent generation

F₁ generation: First filial generation

F₂ generation: Second filial generation

5. Explain how Mendel's simple cross of purple and white flowers did the following:
- a. refuted blending: The reappearance of white-flowered plants in the F₂ generation was evidence that the heritable factor causing white flowers had not been diluted or destroyed by coexisting with the purple-flower factor in the F₁ hybrids.
 - b. determined dominant and recessive characteristics: Mendel reasoned that the heritable factor for white flowers did not disappear in the F₁ plants, but was somehow hidden, or masked, when the purple-flower factor was present. In Mendel's terminology, purple flower color is a dominant trait, and white flower color is a recessive trait.
 - c. demonstrated the merit of experiments that covered multiple generations: Had Mendel stopped his experiments with the F₁ generation, the basic patterns of inheritance would have escaped him. Mendel's quantitative analysis of the F₂ plants from thousands of genetic crosses like these allowed him to deduce two fundamental principles of heredity: the law of segregation and the law of independent assortment.
6. On the figure below, label the *allele* for both purple and white flower color, a *homologous pair*, and the *locus* of the flower color gene.

See page 265 in your text for the labeled figure.

7. In sexually reproducing organisms, why are there exactly two chromosomes in each homologous pair?
- Each somatic cell in a diploid organism has two sets of chromosomes, one set inherited from each parent.
8. Mendel's model consists of four concepts. Describe each concept in the appropriate space below. Indicate which of the concepts can be observed during meiosis by placing an asterisk by the concept.

Mendel's Four Concepts	Description of Concept
First concept	Alternative versions of genes account for variations in inherited characters.
Second concept	For each character, an organism inherits two copies of a gene, one from each parent.

Third concept	If the two alleles at a locus differ, then one, the dominant allele, determines the organism's appearance; the other, the recessive allele, has no noticeable effect on the organism's appearance.
Fourth concept (law of segregation)	The two alleles for a heritable character segregate (separate from each other) during gamete formation and end up in different gametes.*

9. Using Figure 14.5 in your text as your guide, provide the missing notations for the figure below. (P, F₁, F₂). Also indicate the alleles for each individual as well as the gametes it produces, and complete the Punnett square.
- What is the F₂ phenotypic and genotypic ratio? **phenotype 3:1, genotype 1:2:1**
 - Which generation is completely heterozygous? **F₁ generation**
 - Which generation has both heterozygous and homozygous offspring? **F₂ generation**

See page 266 in your text for the labeled figure.

10. In pea plants, *T* is the allele for tall plants, while *t* is the allele for dwarf plants. If you have a tall plant, demonstrate with a *testcross* how it could be determined if the plant is homozygous tall or heterozygous tall.

See page 267 in your text for an example of a testcross.

A testcross always involves crossing the unknown phenotype with an individual that is homozygous recessive for the trait in question. In this case, a homozygous tall plant crossed with a homozygous recessive dwarf will yield all tall offspring. A heterozygous tall plant crossed with a dwarf will yield an offspring ratio of one tall plant to one dwarf plant. The presence of dwarf plants indicates that the previously unknown tall plant is heterozygous.

11. Explain the difference between a *monohybrid* cross and a *dihybrid* cross.

Monohybrid cross: A cross between two organisms that are heterozygous for the character being followed (or the self-pollination of a heterozygous plant)

Dihybrid cross: A cross between two organisms that are each heterozygous for both of the characters being followed (or the self-pollination of a plant that is heterozygous for both characters)

12. As you start to work word problems in genetics, two things are critical: the parent's genotype must be correct, and the gametes must be formed correctly. Using Figure 14.8 as your guide, explain how the gametes are derived for the following cross. (You should have four different gametes).

$$YyRr \times YyRr$$

In our example, an F₁ plant will produce four classes of gametes in equal quantities: YR, Yr, yR, and yr. If sperm of the four classes fertilize eggs of the four classes, there will be 16 (4 × 4) equally probable ways in which the alleles can combine in the F₂ generation, as shown in Figure 14.8, right side. These combinations result in four phenotypic categories with a ratio of 9:3:3:1 (nine yellow-round to three green-round to three yellow-wrinkled to one green-wrinkled).

13. Complete the cross given in question 12 by placing the gametes in a *Punnett square*. Then provide the phenotypic ratio of the offspring.

See page 268 in your text for the labeled figure.

14. Explain Mendel's *law of independent assortment*.

Each pair of alleles segregates independently of each other pair of alleles during gamete formation.

Before leaving this concept, it would be helpful to complete the three problems in *Concept Check 14.1* on page 269 of your textbook. The problems are worked and explained in the Answer section on page A-13 at the back of the book.

Concept 14.2 The laws of probability govern Mendelian inheritance

15. An event that is certain to occur has a probability of **1**, while an event that is certain not to occur has a probability of **0**.

16. In probability, what is an *independent event*?

An event whose outcome is unaffected by what has happened on previous trials, such as in a sequence of coin tosses

17. State the *multiplication rule* and give an original example.

The multiplication rule states that to determine this probability, we multiply the probability of one event by the probability of the other event. For example, by the multiplication rule, the probability that both coins will land heads up is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$.

Original examples will vary.

18. State the *addition rule* and give an original example.

The addition rule states that the probability that any two or more mutually exclusive events will occur is calculated by adding their individual probabilities.

Original examples will vary.

19. What is the probability that a couple will have a girl, a boy, a girl, and a boy in this specific order?

The probability is 1/16.

Concept 14.3 Inheritance patterns are often more complex than those predicted by simple Mendelian genetics

20. Explain how *incomplete dominance* is different from *complete dominance*, and give an example of incomplete dominance.

Incomplete dominance is the situation in which the phenotype of heterozygotes is intermediate between the phenotypes of individuals homozygous for either allele.

Complete dominance is the situation in which the phenotypes of the heterozygote and dominant homozygote are indistinguishable.

An example of incomplete dominance is the crossing of red snapdragons with white snapdragons to produce F₁ hybrids with pink flowers.

21. Compare and contrast *codominance* with *incomplete dominance*.

In codominance, the phenotypes of both alleles are exhibited in the heterozygote because both alleles affect the phenotype in separate, distinguishable ways, such as in the human MN blood group, determined by the codominant alleles for two specific molecules located on the surface of red blood cells, the M and N molecules.

In incomplete dominance, the phenotype of heterozygotes is intermediate between the phenotypes of individuals homozygous for either allele; neither allele is completely dominant, and the F₁ hybrids have a phenotype somewhere between those of the two parental varieties.

22. Dominant alleles are not necessarily more common than recessive alleles in the gene pool. Explain why this is true.

Natural selection determines how common an allele is in the gene pool. For example, having six fingers (polydactyly) is dominant to five fingers, but the presence of six fingers is not common in the human gene pool.

23. Explain what is meant when a gene is said to have *multiple alleles*. Blood groups are an excellent human example of this.

Most genes exist in more than two allelic forms, for example, ABO blood groups.

24. Blood groups are so important medically that you should be able to solve genetics problems based on blood types. The first step in accomplishing that is to understand the genotypes of each blood type. Before working any problems, complete this ABO blood type chart.

Genotype	Red Blood Cell Appearance	Phenotype (blood group)
I ^A I ^A or I ^A i	See Figure 14.11.	A

$I^B I^B$ or $I^B i$	See Figure 14.11.	B
$I^A I^B$	See Figure 14.11.	AB
ii	See Figure 14.11.	O

25. Question 2 in the *Concept Check 14.3* is a blood type problem. Complete it here, and show your work.

Question: If a man with type AB blood marries a woman with type O, what blood types would you expect in their children? What fraction would you expect of each type?

Answer: Half of the children would be expected to have type A blood and half type B blood.

26. What is *pleiotropy*? Explain why this is important in diseases like cystic fibrosis and sickle-cell disease.

Pleiotropy is the ability of a single gene to have multiple effects. In humans, pleiotropic alleles are responsible for multiple symptoms associated with certain hereditary diseases, such as cystic fibrosis and sickle-cell disease.

27. Explain *epistasis*.

Epistasis is a type of gene interaction in which the phenotypic expression of one gene alters that of another independently inherited gene.

28. Explain why the dihybrid cross detailed in Figure 14.12 in your text has four yellow Labrador retrievers instead of the three that would have been predicted by Mendel's work.

This dihybrid cross results in four yellow Labrador retrievers rather than three because the dominant allele, symbolized by E, results in the deposition of either black or brown pigment. If the Lab is homozygous recessive for the second locus (ee), then the coat is yellow, regardless of the genotype at the black/brown locus. The E/e gene is epistatic to the B/b gene.

29. Why is height a good example of *polygenic inheritance*?

For many characters, such as human skin color and height, an either-or classification is impossible because the characters vary in the population in gradients along a continuum.

30. *Quantitative variation* usually indicates *polygenic inheritance*.

31. Using the terms *norm of reaction* and *multifactorial*, explain the potential influence of the environment on phenotypic expression.

The outcome of a genotype lies within its norm of reaction, a phenotype range that depends on the environment in which the genotype is expressed. For some characters, such as the ABO blood group system, the norm of reaction has no breadth whatsoever. Other characteristics, such as a person's blood count of red and white cells, varies quite a bit, depending on such factors as the altitude, the customary level of physical activity, and the presence of infectious agents. Genetics refers to such characters as multifactorial, meaning that many factors, both genetic and environmental, collectively influence phenotype.

Concept 14.4 Many human traits follow Mendelian patterns of inheritance

32. Pedigree analysis is often used to determine the mode of inheritance (dominant or recessive, for example). Be sure to read the "Tips for pedigree analysis" in Figure 14.15 in your text; then complete the unlabeled pedigree by indicating the genotypes for all involved.

See page 276 in your text for the labeled figure.

What is the mode of inheritance for this pedigree? **Recessive**

33. In the pedigree you completed above, explain why you know the genotype of one female in the third generation, but are unsure of the other.

The presence of a free earlobe could indicate either an FF or Ff genotype, as F is the dominant allele, resulting in free earlobes.

The female with the recessive trait can only have one genotype. The female with the dominant trait could be homozygous or heterozygous.

34. Describe what you think is medically important to know about the behavior of recessive alleles.

Thousands of genetic disorders are known to be inherited as simple recessive traits. These disorders range in severity from relatively mild, such as albinism (lack of pigmentation, which results in susceptibility to skin cancers and vision problems) to life-threatening, such as cystic fibrosis.

35. You are expected to have a general knowledge of the pattern of inheritance and the common symptoms of a number of genetic disorders. Provide this information for the disorders listed below.

a. cystic fibrosis: A human genetic disorder caused by a recessive allele for a chloride channel protein; characterized by an excessive secretion of mucus and consequent vulnerability to infection; fatal if untreated.

b. sickle-cell disease: A recessively inherited human blood disorder in which a single nucleotide change in the β -globin gene causes hemoglobin to aggregate, changing red blood cell shape and causing multiple symptoms in afflicted individuals.

c. achondroplasia: A form of dwarfism that occurs in one of every 25,000 people. Heterozygous individuals have the dwarf phenotype. Like the presence of extra fingers or toes, achondroplasia is a trait for which the recessive allele is much more prevalent than the corresponding dominant allele.

d. Huntington's disease: A human genetic disease caused by a dominant allele; characterized by uncontrollable body movements and degeneration of the nervous system; usually fatal 10 to 20 years after the onset of symptoms.

36. *Amniocentesis* and *chorionic villus sampling* are the two most widely used methods for testing a fetus for genetic disorders. Use the unlabeled diagram below to explain the three main steps in amniocentesis and the two main steps of CVS.

See page 281 in your text for the labeled figure.

Amniocentesis:

1. A sample of amniotic fluid can be taken starting at the fourteenth to sixteenth week of pregnancy.
2. Biochemical and genetic tests can be performed immediately on the amniotic fluid or later on the cultured cell.
3. Fetal cells must be cultured for several weeks to obtain sufficient numbers for karyotyping.

CVS:

1. A sample of chorionic villus tissue can be taken as early as the eighth to tenth week of pregnancy.
2. Karyotyping and biochemical and genetic tests can be performed on the fetal cells immediately, providing results within a day or so.

37. What are the strengths and weaknesses of each fetal test?

Strength of amniocentesis: In addition to fetal cells, amniotic fluid is also collected. Amniotic fluid can be used to detect additional enzymatic or developmental problems not detectable from the karyotype.

Weakness of amniocentesis: Cells must be cultured for several weeks before karyotyping, and the test cannot be performed until the fourteenth to sixteenth week.

Strength of CVS: These cells proliferate rapidly enough to allow karyotyping to be carried out immediately, and CVS can be performed as early as the eighth to tenth week.

Weakness of CVS: No amniotic fluid is collected with this technique.

38. What are the symptoms of *phenylketonuria* (PKU)? How is newborn screening used to identify children with this disorder?

The symptoms of phenylketonuria include an inability to metabolize the amino acid phenylalanine, causing severe mental intellectual disability. Some genetic disorders, including phenylketonuria, can be detected at birth by simple biochemical tests that are now routinely performed in most hospitals in the United States.

Test Your Understanding Answers

One of the ways to determine your understanding of Mendelian genetics is to work many genetics problems. Complete the questions for the problems at the end of the chapter.

Before starting, it would be productive to read the “Tips for Genetics Problems” on page 283. Work neatly, and show all work. As you know, you can check your solutions in your text.

[See page A-13 \(Appendix\) of your text for answers.](#)